NEBRASKA HEALTH AND HUMAN SERVICES SYSTEM

ACT SHEET FOR POSTIVE NEWBORN SCREENING RESULT FOR CYSTIC FIBROSIS

Disease Category: Cystic Fibrosis transmembrane conductance regulator (CFTR) deficiency.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and to ascertain clinical status (meconium ileus, failure to thrive, recurrent cough, wheezing and chronic abdominal pain).
- Contact Accredited CF Center for questions, and appointment scheduling.
- Determine sweat chloride (unless two DNA mutations already identified). This may be done by the CF Center.
- If CF is confirmed, clinical evaluation and genetic counseling are indicated.
- Report findings to the newborn screening program.

Accredited CF Center contacts:
Nebraska Accredited CF Center (at UNMC in Omaha): 402 559-6275
Colorado Accredited CF Center (Children's in Denver): 303-861-6182
http://www.cysticfibrosis.com/centers.html

Meaning of the Screening Result:
A positive screening result can be obtained on an initial specimen or on a repeat screen with an elevated immunoreactive trypsinogen (IRT) result in conjunction with two copies of DNA mutations known to be associated with cystic fibrosis.

Condition Description: The CFTR protein regulates chloride transport that is important for function of lungs, upper respiratory tract, pancreas, liver, sweat glands and genitourinary tract.

Confirmation of Diagnosis:
Genotype-phenotype associations are known for many CF mutations and combinations of mutations. Therefore, it may not be necessary to conduct a sweat test for some patients when this information is derived from the screen. However it is important to repeat a dried blood spot filter paper specimen to verify the correct child has been screened and reported. Referral to an accredited CF center is always recommended for comprehensive management of CF patients. Patients with "inconclusive" initial or repeat screening results due to elevated IRT's with a single mutation identified , or repeat screens with continued IRT elevations alone are always recommended for sweat chloride iontophoresis testing for definitive diagnosis.

Clinical Expectations: Deficient chloride transport in lungs leads to production of abnormally thick mucous leading to airway obstruction, neutrophil dominated inflammation and recurrent and progressive pulmonary infections. Pancreatic insufficiency is found in 80-90% of cases.

Additional Information:

- Cystic Fibrosis Foundation - www.cff.org
- New England Metabolic Consortium - www.childrenshospital.org/newenglandconsortium
- Gene Tests/Gene Clinics - www.genetests.org
- U.S. National Newborn Screening & Genetics Resource Center - www.genes-r-us.uthscsa.edu